Heidi L Rehm

List of Publications by Year
in descending order

244 papers	45,081 citations	10986 71 h-index	2385 198 g-index
286 all docs	286 docs citations	286 times ranked	48929 citing authors

#	Article	IF	Citations
1	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine, 2015, 17, 405-424.	2.4	20,455
2	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genetics in Medicine, 2013, 15, 565-574.	2.4	2,186
3	Guidelines for investigating causality of sequence variants in human disease. Nature, 2014, 508, 469-476.	27.8	1,130
4	ClinGen â€" The Clinical Genome Resource. New England Journal of Medicine, 2015, 372, 2235-2242.	27.0	1,016
5	ACMG clinical laboratory standards for next-generation sequencing. Genetics in Medicine, 2013, 15, 733-747.	2.4	794
6	TRPA1 is a candidate for the mechanosensitive transduction channel of vertebrate hair cells. Nature, 2004, 432, 723-730.	27.8	657
7	Genetic Misdiagnoses and the Potential for Health Disparities. New England Journal of Medicine, 2016, 375, 655-665.	27.0	602
8	Recommendations for interpreting the loss of function PVS1 ACMG/AMP variant criterion. Human Mutation, 2018, 39, 1517-1524.	2.5	511
9	GJB2 Mutations and Degree of Hearing Loss: A Multicenter Study. American Journal of Human Genetics, 2005, 77, 945-957.	6.2	455
10	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	27.8	441
11	Assuring the quality of next-generation sequencing in clinical laboratory practice. Nature Biotechnology, 2012, 30, 1033-1036.	17.5	437
12	Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium. American Journal of Human Genetics, 2016, 98, 1067-1076.	6.2	432
13	Standardizing terms for clinical pharmacogenetic test results: consensus terms from the Clinical Pharmacogenetics Implementation Consortium (CPIC). Genetics in Medicine, 2017, 19, 215-223.	2.4	410
14	Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource. American Journal of Human Genetics, 2017, 100, 895-906.	6.2	403
15	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	2.5	390
16	Building the foundation for genomics in precision medicine. Nature, 2015, 526, 336-342.	27.8	376
17	Mutation of a Gene Encoding a Protein with Extracellular Matrix Motifs in Usher Syndrome Type IIa. Science, 1998, 280, 1753-1757.	12.6	366
18	Shared Genetic Causes of Cardiac Hypertrophy in Children and Adults. New England Journal of Medicine, 2008, 358, 1899-1908.	27.0	352

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19	Disease-targeted sequencing: a cornerstone in the clinic. Nature Reviews Genetics, 2013, 14, 295-300.	16.3	349
20	Results of clinical genetic testing of 2,912 probands with hypertrophic cardiomyopathy: expanded panels offer limited additional sensitivity. Genetics in Medicine, 2015, 17, 880-888.	2.4	344
21	Return of Genomic Results to Research Participants: The Floor, the Ceiling, and the Choices In Between. American Journal of Human Genetics, 2014, 94, 818-826.	6.2	342
22	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	5.5	313
23	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	2.5	312
24	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
25	The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. Genetics in Medicine, 2014, 16, 601-608.	2.4	284
26	Lack Of Diversity In Genomic Databases Is A Barrier To Translating Precision Medicine Research Into Practice. Health Affairs, 2018, 37, 780-785.	5.2	213
27	American College of Medical Genetics and Genomics guideline for the clinical evaluation and etiologic diagnosis of hearing loss. Genetics in Medicine, 2014, 16, 347-355.	2.4	207
28	How many rare diseases are there?. Nature Reviews Drug Discovery, 2020, 19, 77-78.	46.4	204
29	Clinical laboratories collaborate to resolve differences in variant interpretations submitted to ClinVar. Genetics in Medicine, 2017, 19, 1096-1104.	2.4	200
30	A public resource facilitating clinical use of genomes. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 11920-11927.	7.1	194
31	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4667-4672.	7.1	193
32	Variant interpretation using population databases: Lessons from gnomAD. Human Mutation, 2022, 43, 1012-1030.	2.5	184
33	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	6.2	176
34	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	2.1	174
35	Inherited Cardiomyopathies. Journal of Molecular Diagnostics, 2013, 15, 158-170.	2.8	172
36	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161

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37	Exploring concordance and discordance for return of incidental findings from clinical sequencing. Genetics in Medicine, 2012, 14, 405-410.	2.4	149
38	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	3.5	148
39	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 2348-2361.	6.1	147
40	Global implementation of genomic medicine: We are not alone. Science Translational Medicine, 2015, 7, 290ps13.	12.4	146
41	Genetic Testing for Dilated Cardiomyopathy in Clinical Practice. Journal of Cardiac Failure, 2012, 18, 296-303.	1.7	145
42	The Impact of Whole-Genome Sequencing on the Primary Care and Outcomes of Healthy Adult Patients. Annals of Internal Medicine, 2017, 167, 159.	3.9	145
43	Phased Whole-Genome Genetic Risk in a Family Quartet Using a Major Allele Reference Sequence. PLoS Genetics, 2011, 7, e1002280.	3.5	137
44	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
45	Vascular Defects and Sensorineural Deafness in a Mouse Model of Norrie Disease. Journal of Neuroscience, 2002, 22, 4286-4292.	3.6	136
46	A multicenter study of the frequency and distribution of GJB2 and GJB6 mutations in a large North American cohort. Genetics in Medicine, 2007, 9, 413-426.	2.4	134
47	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
48	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and geneâ€level specification of the ACMG/AMP guidelines for sequence variant interpretation. Human Mutation, 2018, 39, 1614-1622.	2.5	132
49	Connexin 26 Studies in Patients With Sensorineural Hearing Loss. JAMA Otolaryngology, 2001, 127, 1037.	1.2	126
50	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
51	The MedSeq Project: a randomized trial of integrating whole genome sequencing into clinical medicine. Trials, 2014, 15, 85.	1.6	122
52	Burden of Rare Sarcomere Gene Variants in the Framingham and Jackson Heart Study Cohorts. American Journal of Human Genetics, 2012, 91, 513-519.	6.2	116
53	New Approaches to Molecular Diagnosis. JAMA - Journal of the American Medical Association, 2013, 309, 1511.	7.4	116
54	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	1.7	115

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55	Secondary findings from clinical genomic sequencing: prevalence, patient perspectives, family history assessment, and health-care costs from a multisite study. Genetics in Medicine, 2019, 21, 1100-1110.	2.4	111
56	Research Directions in the Clinical Implementation of Pharmacogenomics: An Overview of US Programs and Projects. Clinical Pharmacology and Therapeutics, 2018, 103, 778-786.	4.7	110
57	Problems with Using Polygenic Scores to Select Embryos. New England Journal of Medicine, 2021, 385, 78-86.	27.0	105
58	Updated recommendation for the benign standâ€alone ACMG/AMP criterion. Human Mutation, 2018, 39, 1525-1530.	2.5	102
59	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
60	Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequence data in the Clinical Sequencing Exploratory Research Consortium. Genetics in Medicine, 2013, 15, 860-867.	2.4	99
61	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605.	6.2	99
62	Evolving health care through personal genomics. Nature Reviews Genetics, 2017, 18, 259-267.	16.3	98
63	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
64	The Responsibility to Recontact Research Participants after Reinterpretation of Genetic and Genomic Research Results. American Journal of Human Genetics, 2019, 104, 578-595.	6.2	91
65	Using ClinVar as a Resource to Support Variant Interpretation. Current Protocols in Human Genetics, 2016, 89, 8.16.1-8.16.23.	3.5	89
66	Overview of Specifications to the ACMG/AMP Variant Interpretation Guidelines. Current Protocols in Human Genetics, 2019, 103, e93.	3.5	88
67	Communicating new knowledge on previously reported genetic variants. Genetics in Medicine, 2012, 14, 713-719.	2.4	87
68	Audiologic Phenotype and Progression in GJB2 (Connexin 26) Hearing Loss. JAMA Otolaryngology, 2010, 136, 81.	1.2	84
69	A systematic approach to the reporting of medically relevant findings from whole genome sequencing. BMC Medical Genetics, 2014, 15, 134.	2.1	84
70	Disease Boundaries in the Retina of Patients with Usher Syndrome Caused byMYO7AGene Mutations. , 2009, 50, 1886.		83
71	Association of Racial/Ethnic Categories With the Ability of Genetic Tests to Detect a Cause of Cardiomyopathy. JAMA Cardiology, 2018, 3, 341.	6.1	83
72	Allelic hierarchy of CDH23 mutations causing non-syndromic deafness DFNB12 or Usher syndrome USH1D in compound heterozygotes. Journal of Medical Genetics, 2011, 48, 767-775.	3.2	82

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73	Comprehensive red blood cell and platelet antigen prediction from whole genome sequencing: proof of principle. Transfusion, 2016, 56, 743-754.	1.6	81
74	ClinVar Miner: Demonstrating utility of a Web-based tool for viewing and filtering ClinVar data. Human Mutation, 2018, 39, 1051-1060.	2.5	81
75	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	2.4	79
76	Genomeâ€wide SNP genotyping identifies the Stereocilin (STRC) gene as a major contributor to pediatric bilateral sensorineural hearing impairment. American Journal of Medical Genetics, Part A, 2012, 158A, 298-308.	1.2	78
77	Is â€˜likely pathogenicâ€™ really 90% likely? Reclassification data in ClinVar. Genome Medicine, 2019, 11, 72.	8.2	78
78	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
79	The GeneInsight suite: a platform to support laboratory and provider use of DNA-based genetic testing. Human Mutation, 2011, 32, 532-536.	2.5	75
80	Development and Validation of a Computational Method for Assessment of Missense Variants in Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 183-192.	6.2	73
81	A novel custom resequencing array for dilated cardiomyopathy. Genetics in Medicine, 2010, 12, 268-278.	2.4	71
82	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. Lancet Haematology,the, 2018, 5, e241-e251.	4.6	70
83	Variant Interpretation for Dilated Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, e002480.	3.6	70
84	Retinal Disease Course in Usher Syndrome 1B Due toMYO7AMutations. , 2011, 52, 7924.		68
85	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	2.4	68
86	Short Communication: The Cardiac Myosin Binding Protein C Arg502Trp Mutation. Circulation Research, 2010, 106, 1549-1552.	4.5	67
87	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
88	Consent Codes: Upholding Standard Data Use Conditions. PLoS Genetics, 2016, 12, e1005772.	3.5	65
89	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	8.2	65
90	Next-generation sequencing for constitutional variants in the clinical laboratory, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1399-1415.	2.4	64

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91	Improving hearing loss gene testing: a systematic review of gene evidence toward more efficient next-generation sequencingâ€“based diagnostic testing and interpretation. Genetics in Medicine, 2016, 18, 545-553.	2.4	63
92	A Comparison of Whole Genome Sequencing to Multigene Panel Testing in Hypertrophic Cardiomyopathy Patients. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	62
93	VisCap: inference and visualization of germ-line copy-number variants from targeted clinical sequencing data. Genetics in Medicine, 2016, 18, 712-719.	2.4	61
94	Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477.	2.4	61
95	ClinGen Pathogenicity Calculator: a configurable system for assessing pathogenicity of genetic variants. Genome Medicine, 2017, 9, 3.	8.2	59
96	Mitochondrial DNA variation across 56,434 individuals in gnomAD. Genome Research, 2022, 32, 569-582.	5.5	59
97	Filter-based hybridization capture of subgenomes enables resequencing and copy-number detection. Nature Methods, 2009, 6, 507-510.	19.0	56
98	GenomeConnect: Matchmaking Between Patients, Clinical Laboratories, and Researchers to Improve Genomic Knowledge. Human Mutation, 2015, 36, 974-978.	2.5	56
99	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genetics in Medicine, 2019, 21, 2442-2452.	2.4	56
100	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
101	Aggregate penetrance of genomic variants for actionable disorders in European and African Americans. Science Translational Medicine, 2016, 8, 364ra151.	12.4	55
102	Comprehensive Diagnostic Testing for Stereocilin. Journal of Molecular Diagnostics, 2014, 16, 639-647.	2.8	53
103	Variant Classification Concordance using the ACMG-AMP Variant Interpretation Guidelines across Nine Genomic Implementation Research Studies. American Journal of Human Genetics, 2020, 107, 932-941.	6.2	51
104	Scaling resolution of variant classification differences in ClinVar between 41 clinical laboratories through an outlier approach. Human Mutation, 2018, 39, 1641-1649.	2.5	50
105	A Rigorous Interlaboratory Examination of the Need to Confirm Next-Generation Sequencingâ€“Detected Variants with an Orthogonal MethodÂin Clinical Genetic Testing. Journal of Molecular Diagnostics, 2019, 21, 318-329.	2.8	49
106	Best practices for the interpretation and reporting of clinical whole genome sequencing. Npj Genomic Medicine, 2022, 7, 27.	3.8	48
107	High-throughput detection of mutations responsible for childhood hearing loss using resequencing microarrays. BMC Biotechnology, 2010, 10, 10.	3.3	47
108	Matchmaker Exchange. Current Protocols in Human Genetics, 2017, 95, 9.31.1-9.31.15.	3.5	47

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109	Prenatal DNA Sequencing: Clinical, Counseling, and Diagnostic Laboratory Considerations. Prenatal Diagnosis, 2018, 38, 26-32.	2.3	47
110	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	2.4	47
111	Norrie disease: Extraocular clinical manifestations in 56 patients. American Journal of Medical Genetics, Part A, 2012, 158A, 1909-1917.	1.2	45
112	All the World's a Stage: Facilitating Discovery Science and Improved Cancer Care through the Global Alliance for Genomics and Health. Cancer Discovery, 2015, 5, 1133-1136.	9.4	45
113	Returning a Genomic Result for an Adult-Onset Condition to the Parents of a Newborn: Insights From the BabySeq Project. Pediatrics, 2019, 143, S37-S43.	2.1	45
114	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. Genetics in Medicine, 2019, 21, 2496-2503.	2.4	45
115	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
116	Implications of Hypertrophic Cardiomyopathy Transmitted by Sperm Donation. JAMA - Journal of the American Medical Association, 2009, 302, 1681.	7.4	43
117	Evaluation of Second-Generation Sequencing of 19 Dilated Cardiomyopathy Genes for Clinical Applications. Journal of Molecular Diagnostics, 2010, 12, 818-827.	2.8	43
118	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. Genetics in Medicine, 2017, 19, 1245-1252.	2.4	43
119	GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2123-2138.	6.1	42
120	Toward Genetics-Driven Early Intervention in Dilated Cardiomyopathy. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	41
121	Use and interpretation of genetic tests in cardiovascular genetics. Heart, 2010, 96, 1669-1675.	2.9	40
122	The Medical Genome Initiative: moving whole-genome sequencing for rare disease diagnosis to the clinic. Genome Medicine, 2020, 12, 48.	8.2	40
123	Analyzing and Reanalyzing the Genome: Findings from the MedSeq Project. American Journal of Human Genetics, 2019, 105, 177-188.	6.2	38
124	Exome sequencing in infants with congenital hearing impairment: a population-based cohort study. European Journal of Human Genetics, 2020, 28, 587-596.	2.8	38
125	A multicenter study of the frequency and distribution of GJB2 and GJB6 mutations in a large North American cohort. Genetics in Medicine, 2007, 9, 413-26.	2.4	38
126	A One-Page Summary Report of Genome Sequencing for the Healthy Adult. Public Health Genomics, 2015, 18, 123-129.	1.0	37

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127	Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. Human Mutation, 2016, 37, 119-126.	2.5	37
128	MutaDATABASE: a centralized and standardized DNA variation database. Nature Biotechnology, 2011, 29, 117-118.	17.5	36
129	Creating a data resource: what will it take to build a medical information commons?. Genome Medicine, 2017, 9, 84.	8.2	36
130	A survey assessing adoption of the ACMG-AMP guidelines for interpreting sequence variants and identification of areas for continued improvement. Genetics in Medicine, 2019, 21, 1699-1701.	2.4	35
131	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	6.2	35
132	Summarizing polygenic risks for complex diseases in a clinical whole-genome report. Genetics in Medicine, 2015, 17, 536-544.	2.4	34
133	A new era in the interpretation of human genomic variation. Genetics in Medicine, 2017, 19, 1092-1095.	2.4	34
134	Returning actionable genomic results in a research biobank: Analytic validity, clinical implementation, and resource utilization. American Journal of Human Genetics, 2021, 108, 2224-2237.	6.2	34
135	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. Genome Medicine, 2022, 14, 6.	8.2	34
136	Norrie disease gene mutation in a large Costa Rican kindred with a novel phenotype including venous insufficiency. Human Mutation, 1997, 9, 402-408.	2.5	33
137	Registered access: authorizing data access. European Journal of Human Genetics, 2018, 26, 1721-1731.	2.8	33
138	Distinguishing Variant Pathogenicity From Genetic Diagnosis. JAMA - Journal of the American Medical Association, 2018, 320, 1929.	7.4	32
139	Audiologic Features of Norrie Disease. Annals of Otology, Rhinology and Laryngology, 2005, 114, 533-538.	1.1	31
140	seqr : A webâ€based analysis and collaboration tool for rare disease genomics. Human Mutation, 2022, , .	2.5	31
141	Usability of a novel clinician interface for genetic results. Journal of Biomedical Informatics, 2012, 45, 950-957.	4.3	29
142	Clinical Genome Sequencing. , 2013, , 102-122.		29
143	Management of Secondary Genomic Findings. American Journal of Human Genetics, 2020, 107, 3-14.	6.2	29
144	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). Genetics in Medicine, 2018, 20, 294-302.	2.4	27

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145	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. Journal of the American College of Cardiology, 2019, 74, 2623-2634.	2.8	27
146	Genetic variation in the Middle Eastâ€"an opportunity to advance the human genetics field. Genome Medicine, 2020, 12, 116.	8.2	27
147	Whole-genome sequencing as an investigational device for return of hereditary disease risk and pharmacogenomic results as part of the All of Us Research Program. Genome Medicine, 2022, 14, 34.	8.2	27
148	An Overview of Custom Array Sequencing. Current Protocols in Human Genetics, 2009, 61, Unit 7.17.	3.5	25
149	Development and Validation of a Mass Spectrometryâ€"Based Assay for the Molecular Diagnosis of Mucin-1 Kidney Disease. Journal of Molecular Diagnostics, 2016, 18, 566-571.	2.8	25
150	Short-term costs of integrating whole-genome sequencing into primary care and cardiology settings: a pilot randomized trial. Genetics in Medicine, 2018, 20, 1544-1553.	2.4	25
151	ClinGen's GenomeConnect registry enables patientâ€centered data sharing. Human Mutation, 2018, 39, 1668-1676.	2.5	25
152	Curating Clinically Relevant Transcripts for the Interpretation of Sequence Variants. Journal of Molecular Diagnostics, 2018, 20, 789-801.	2.8	25
153	TBC1D8B Mutations Implicate RAB11-Dependent Vesicular Trafficking in the Pathogenesis of Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2019, 30, 2338-2353.	6.1	25
154	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
155	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncology, 2022, 8, 835.	7.1	25
156	A Genetic Approach to the Child with Sensorineural Hearing Loss. Seminars in Perinatology, 2005, 29, 173-181.	2.5	24
157	Allele-Specific Droplet Digital PCR Combined with a Next-Generation Sequencing-Based Algorithm for Diagnostic Copy Number Analysis in Genes with High Homology: Proof of Concept Using Stereocilin. Clinical Chemistry, 2018, 64, 705-714.	3.2	24
158	Points to consider for sharing variant-level information from clinical genetic testing with ClinVar. Journal of Physical Education and Sports Management, 2018, 4, a002345.	1.2	23
159	Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. Frontiers in Genetics, 2021, 12, 674295.	2.3	23
160	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. Genetics in Medicine, 2018, 20, 855-866.	2.4	22
161	International federation of genomic medicine databases using GA4GH standards. Cell Genomics, 2021, 1, 100032.	6.5	22
162	Additional clinical manifestations in children with sensorineural hearing loss and biallelicGJB2 mutations: Who should be offeredGJB2 testing?. American Journal of Medical Genetics, Part A, 2007, 143A, 1560-1566.	1.2	21

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163	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	2.5	20
164	Development of a consent resource for genomic data sharing in the clinical setting. Genetics in Medicine, 2019, 21, 81-88.	2.4	20
165	Utilizing ClinGen geneâ€disease validity and dosage sensitivity curations to inform variant classification. Human Mutation, 2022, 43, 1031-1040.	2.5	20
166	Evaluating the impact of in silico predictors on clinical variant classification. Genetics in Medicine, 2022, 24, 924-930.	2.4	20
167	Molecular Diagnosis of Hearing Loss. Current Protocols in Human Genetics, 2012, 72, Unit 9.16.	3.5	19
168	Principles and Recommendations for Standardizing the Use of the Next-Generation Sequencing Variant File in Clinical Settings. Journal of Molecular Diagnostics, 2017, 19, 417-426.	2.8	19
169	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. Brain, 2021, 144, 2659-2669.	7.6	19
170	Time to make rare disease diagnosis accessible to all. Nature Medicine, 2022, 28, 241-242.	30.7	19
171	Temporal bone abnormalities in children with GJB2 mutations. Laryngoscope, 2011, 121, 630-635.	2.0	18
172	Recurrent variants in OTOF are significant contributors to prelingual nonsydromic hearing loss in Saudi patients. Genetics in Medicine, 2018, 20, 536-544.	2.4	18
173	From Theory to Reality: Establishing a Successful Kidney Genetics Clinic in the Outpatient Setting. Kidney360, 2020, 1, 1099-1106.	2.1	18
174	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
175	The GA4GH Variation Representation Specification: A computational framework for variation representation and federated identification. Cell Genomics, 2021, 1, 100027.	6.5	18
176	Clinical evaluation and etiologic diagnosis of hearing loss: A clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2022, 24, 1392-1406.	2.4	18
177	Genetics and the Genome Project. Ear and Hearing, 2003, 24, 270-274.	2.1	17
178	Homozygosity for the V37I GJB2 mutation in fifteen probands with mild to moderate sensorineural hearing impairment: Further confirmation of pathogenicity and haplotype analysis in Asian populations. American Journal of Medical Genetics, Part A, 2013, 161, 2148-2157.	1.2	17
179	A novel clinician interface to improve clinician access to up-to-date genetic results. Journal of the American Medical Informatics Association: JAMIA, 2014, 21, e117-e121.	4.4	17
180	CDH23 Related Hearing Loss. Otology and Neurotology, 2016, 37, 1583-1588.	1.3	17

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181	Development of Clinical Domain Working Groups for the Clinical Genome Resource (ClinGen): lessons learned and plans for the future. Genetics in Medicine, 2019, 21, 987-993.	2.4	17
182	Randomized prospective evaluation of genome sequencing versus standard-of-care as a first molecular diagnostic test. Genetics in Medicine, 2021, 23, 1689-1696.	2.4	17
183	A protocol for whole-exome sequencing in newborns with congenital deafness: a prospective population-based cohort. BMJ Paediatrics Open, 2017, 1, e000119.	1.4	16
184	Diagnoses of uncertain significance: kidney genetics in the 21st century. Nature Reviews Nephrology, 2020, 16, 616-618.	9.6	16
185	Universal newborn genetic screening for pediatric cancer predisposition syndromes: model-based insights. Genetics in Medicine, 2021, 23, 1366-1371.	2.4	16
186	The Changing Landscape of Molecular Diagnostic Testing: Implications for Academic Medical Centers. Journal of Personalized Medicine, 2016, 6, 8.	2.5	15
187	Health Care Infrastructure for Financially Sustainable Clinical Genomics. Journal of Molecular Diagnostics, 2016, 18, 697-706.	2.8	15
188	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. Genetics in Medicine, 2017, 19, 496-504.	2.4	15
189	ClinGen advancing genomic dataâ€sharing standards as a GA4GH driver project. Human Mutation, 2018, 39, 1686-1689.	2.5	15
190	Approaches to carrier testing and results disclosure in translational genomics research: The clinical sequencing exploratory research consortium experience. Molecular Genetics & Genomic Medicine, 2018, 6, 898-909.	1.2	15
191	Genomic considerations for FHIRÂ®; eMERGE implementation lessons. Journal of Biomedical Informatics, 2021, 118, 103795.	4.3	15
192	ClinGen and ClinVar â€“ Enabling Genomics in Precision Medicine. Human Mutation, 2018, 39, 1473-1475.	2.5	14
193	Misattributed parentage as an unanticipated finding during exome/genome sequencing: current clinical laboratory practices and an opportunity for standardization. Genetics in Medicine, 2019, 21, 861-866.	2.4	14
194	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. American Journal of Human Genetics, 2021, 108, 357-367.	6.2	14
195	Monogenic and Polygenic Contributions to QTc Prolongation in the Population. Circulation, 2022, 145, 1524-1533.	1.6	14
196	A gene-to-patient approach uplifts novel disease gene discovery and identifies 18 putative novel disease genes. Genetics in Medicine, 2022, 24, 1697-1707.	2.4	14
197	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	2.5	13
198	Professional responsibilities regarding the provision, publication, and dissemination of patient phenotypes in the context of clinical genetic and genomic testing: points to considerâ€”a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 169-171.	2.4	13

#	Article	IF	Citations
199	Introduction of genomics into prenatal diagnostics. Lancet, The, 2019, 393, 719-721.	13.7	13
200	A whole genome approach for discovering the genetic basis of blood group antigens: independent confirmation for P1 and Xg^a. Transfusion, 2019, 59, 908-915.	1.6	13
201	Keeping up with the genomes: scaling genomic variant interpretation. Genome Medicine, 2020, 12, 5.	8.2	13
202	KCND2 variants associated with global developmental delay differentially impair Kv4.2 channel gating. Human Molecular Genetics, 2021, 30, 2300-2314.	2.9	12
203	JAK inhibition in a patient with a STAT1 gain-of-function variant reveals STAT1 dysregulation as a common feature of aplastic anemia. Med, 2022, 3, 42-57.e5.	4.4	11
204	Lumping versus splitting: How to approach defining a disease to enable accurate genomic curation. Cell Genomics, 2022, 2, 100131.	6.5	11
205	Seven years since the launch of the Matchmaker Exchange: The evolution of genomic matchmaking. Human Mutation, 2022, 43, 659-667.	2.5	11
206	ThePTPN11gene is not implicated in nonsyndromic hypertrophic cardiomyopathy. , 2005, 132A, 333-334.		10
207	Targeted Sequencing Using Affymetrix CustomSeq Arrays. Current Protocols in Human Genetics, 2011, 69, Unit7.18.	3.5	10
208	Analysis of two Arab families reveals additional support for a DFNB2 nonsyndromic phenotype of MYO7A. Molecular Biology Reports, 2014, 41, 193-200.	2.3	10
209	Evaluation: A Qualitative Pilot Study of Novel Information Technology Infrastructure to Communicate Genetic Variant Updates. Applied Clinical Informatics, 2016, 07, 461-476.	1.7	10
210	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	2.5	10
211	TMPRSS3 Gene Variants With Implications for Auditory Treatment and Counseling. Frontiers in Genetics, 2021, 12, 780874.	2.3	10
212	Analysis of intragenic USH2A copy number variation unveils broad spectrum of unique and recurrent variants. European Journal of Medical Genetics, 2018, 61, 621-626.	1.3	9
213	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. European Journal of Human Genetics, 2021, 29, 988-997.	2.8	8
214	Information Technology Support for Clinical Genetic Testing within an Academic Medical Center. Journal of Personalized Medicine, 2016, 6, 4.	2.5	7
215	Response to Biesecker and Harrison. Genetics in Medicine, 2018, 20, 1689-1690.	2.4	7
216	The Ancestral Pace of Variant Reclassification. Journal of the National Cancer Institute, 2018, 110, 1133-1134.	6.3	7

#	Article	IF	Citations
217	Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	1.2	7
218	Recontacting registry participants with genetic updates through GenomeConnect, the ClinGen patient registry. Genetics in Medicine, 2021, 23, 1738-1745.	2.4	7
219	Rare Coding Variants Associated With Electrocardiographic Intervals Identify Monogenic Arrhythmia Susceptibility Genes: A Multi-Ancestry Analysis. Circulation Genomic and Precision Medicine, 2021, 14, e003300.	3.6	7
220	LB-11. Comparison of Viral Loads in Individuals With or Without Symptoms At Time of COVID-19 Testing Among 32,480 Residents and Staff of Nursing Homes and Assisted Living Facilities in Massachusetts. Open Forum Infectious Diseases, 2020, 7, S848-S849.	0.9	7
221	Harmonizing variant classification for return of results in the All of Us Research Program. Human Mutation, 2022, 43, 1114-1121.	2.5	7
222	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. Value in Health, 2020, 23, 559-565.	0.3	6
223	The intersection of genetics and COVID-19 in 2021: preview of the 2021 Rodney Howell Symposium. Genetics in Medicine, 2021, 23, 1001-1003.	2.4	6
224	Exome survey of individuals affected by VATER / VACTERL with renal phenotypes identifies phenocopies and novel candidate genes. American Journal of Medical Genetics, Part A, 2021, 185, 3784-3792.	1.2	6
225	Reanalysis of eMERGE phase III sequence variants in 10,500 participants and infrastructure to support the automated return of knowledge updates. Genetics in Medicine, 2022, 24, 454-462.	2.4	6
226	PECONPI: A novel software for uncovering pathogenic copy number variations in nonâ€syndromic sensorineural hearing loss and other genetically heterogeneous disorders. American Journal of Medical Genetics, Part A, 2013, 161, 2134-2147.	1.2	5
227	Reclassification of the BRAF p.Ile208Val variant by case-level data sharing. Journal of Physical Education and Sports Management, 2018, 4, a002675.	1.2	4
228	Rates of Actionable Genetic Findings in Individuals with Colorectal Cancer or Polyps Ascertained from a Community Medical Setting. American Journal of Human Genetics, 2019, 105, 526-533.	6.2	4
229	Primary care providersâ€™ responses to unsolicited Lynch syndrome secondary findings of varying clinical significance. Genetics in Medicine, 2021, 23, 1977-1983.	2.4	4
230	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	2.4	3
231	Rapid communication of efforts to resolve differences or update variant interpretations in ClinVar through case-level data sharing. Journal of Physical Education and Sports Management, 2018, 4, a003467.	1.2	2
232	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471.	0.8	2
233	Creation of an Expert Curated Variant List for Clinical Genomic Test Development and Validation. Journal of Molecular Diagnostics, 2021, 23, 1500-1505.	2.8	2
234	Early childhood hearing loss: Clinical and molecular genetics. An educational slide set of the American College of Medical Genetics. Genetics in Medicine, 2003, 5, 338-341.	2.4	2

#	Article	IF	Citations
235	Correspondence on â€œThe role of clinical response to treatment in determining pathogenicity of genomic variantsâ€•by Shen et al.. Genetics in Medicine, 2021, 23, 586.	2.4	1
236	An Investigation of the Knowledge Overlap between Pharmacogenomics and Disease Genetics. , 2021, , .		1
237	An Investigation of the Knowledge Overlap between Pharmacogenomics and Disease Genetics. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2022, 27, 385-396.	0.7	1
238	Molecular Diagnosis of Hearing Loss. Current Protocols in Human Genetics, 2004, 43, Unit9.16.	3.5	0
239	Translating Research Discoveries Into Clinical Tests. Seminars in Nephrology, 2010, 30, 426-430.	1.6	0
240	Reply to Clarity and claims in variation/mutation databasing. Nature Biotechnology, 2011, 29, 792-794.	17.5	0
241	Rapid access to genetic discoveries underlying human disease. Journal of Physical Education and Sports Management, 2016, 2, a001545.	1.2	0
242	Peter Bauer, Ellen Karges, Gabriela Oprea and Arndt Rolfs. Genetics in Medicine, 2018, 20, 378-379.	2.4	0
243	The Lifespan of Genetic Testing. American Journal of Medicine, 2018, 131, 991-992.	1.5	0
244	Abstract 1192: The Clinical Genome Resource (ClinGen) somatic cancer clinical domain working group. Cancer Research, 2022, 82, 1192-1192.	0.9	0